GPR143 gene

G protein-coupled receptor 143

Normal Function

The *GPR143* gene, also known as *OA1*, provides instructions for making a protein that is involved in the coloring (pigmentation) of the eyes and skin. This protein is made in the light-sensitive tissue at the back of the eye (the retina) and in skin cells. The GPR143 protein is part of a signaling pathway that controls the growth and maturation of melanosomes, which are cellular structures that produce and store a pigment called melanin. Melanin is the substance that gives skin, hair, and eyes their color. In the retina, this pigment also plays a critical role in normal vision.

Health Conditions Related to Genetic Changes

ocular albinism

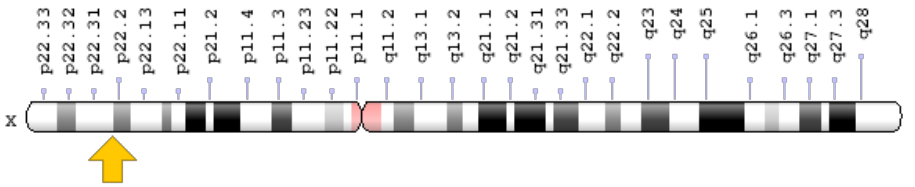
More than 60 *GPR143* mutations have been identified in people with the most common form of ocular albinism, which is called the Nettleship-Falls type or type 1. Most mutations alter the size or shape of the GPR143 protein. These genetic changes often prevent the abnormal protein from ever reaching melanosomes, where it is needed to control the growth of these pigment-containing structures. In other cases, the GPR143 protein reaches melanosomes normally but mutations prevent the protein from interacting with other molecules in its signaling pathway. Without functional GPR143 protein, melanosomes in skin cells and the retina can grow abnormally large. It is unclear how these giant melanosomes (macromelanosomes) are related to vision loss and other eye abnormalities in people with ocular albinism.

Most forms of albinism result from a reduced amount of melanin pigment within cells. Researchers continue to study why ocular albinism occurs when cells in the retina appear to contain a substantial amount of melanin. It is possible that this pigment is concentrated into a few abnormal macromelanosomes instead of being evenly distributed among many normal-sized melanosomes within the cell. Additional studies may help clarify the relationship between melanosomes, melanin distribution, and the reduced levels of pigmentation that are characteristic of ocular albinism.

Chromosomal Location

Cytogenetic Location: Xp22.2, which is the short (p) arm of the X chromosome at position 22.2

Molecular Location: base pairs 9,725,413 to 9,765,965 on the X chromosome (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- GP143_HUMAN
- OA1
- ocular albinism 1 (Nettleship-Falls)
- ocular albinism type 1 protein

Additional Information & Resources

GeneReviews

- Ocular Albinism, X-Linked
  https://www.ncbi.nlm.nih.gov/books/NBK1343

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28GPR143%5BTIAB%5D%29+OR+%28%28OA1%5BTIAB%5D%29+OR+%28ocular+albinism+1%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2160+days%22%5Bdp%5D

OMIM

- G PROTEIN-COUPLED RECEPTOR 143
  http://omim.org/entry/300808

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_GPR143.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=GPR143%5Bgene%5D
- HGNC Gene Family: 7TM orphan receptors
  http://www.genenames.org/cgi-bin/genefamilies/set/310
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=20145
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/4935
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  http://www.uniprot.org/uniprot/P51810

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